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Case Report

What is in the rectum? A rare case of gastric heterotopia

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ABSTRACT

Gastric heterotopia (GH) is the occurrence of normal gastric tissue outside of its typical location in the stomach. Although it has been documented in various locations, its presence in the rectum is particularly rare. Due to a lack of available evidence, the exact pathophysiology, malignant potential and appropriate surveillance of GH in the rectum is unclear, thus creating challenges for clinicians managing these patients. This case presents at 46-year-old male who was incidentally found to have GH in the rectum.

Keywords: Gastric heterotopia, Rectum

INTRODUCTION

Gastric heterotopia (GH) refers to the presence of normal gastric mucosa in an abnormal location.[1] It has been described in various sites of the gastrointestinal tract with it most frequently seen in the oesophagus, duodenum or Meckel's diverticulum.[1] It's occurrence in the anorectal region however is exceedingly rare with fewer than 80 cases reported in the literature since it was first described by Ewell and Jackson in 1939.[2] Accurate diagnosis of GH is made through histopathological analysis as endoscopically it can be mistaken for diverticulum, ulceration or malignancy.[3]

CASE REPORT

A 46-year-old male was referred for an outpatient colonoscopy following a positive faecal occult blood test (FOBT). The patient denied any rectal bleeding, perianal pain or bowel symptoms. There was no family history of colorectal cancer or inflammatory bowel disease. His surgical history included a laparoscopic fundoplication in 2018 for gastro-oesophageal reflux disease. He has no other significant medical history. The colonoscopy identified a 2 cm non-bleeding, non-circumferential mass in the distal rectum, located approximately 2-4 cm from the anal verge (Figure 1).

A biopsy was sent for histopathological analysis. The colonoscopy was completed to the terminal ileum and showed sigmoid diverticulosis but was otherwise unremarkable. An upper endoscopy was performed on the same day. Histopathology revealed features of gastric body-type mucosa without evidence of dysplasia or malignancy. The case was discussed at the multidisciplinary team meeting with a recommendation given to proceed with further biopsies of the rectal lesion. Flexible sigmoidoscopy and repeat biopsy revealed gastric foveolar epithelium and associated oxyntic glands without evidence of dysplasia or malignancy, consistent with GH (Figure 2). Endoscopy and histopathology results were explained to the patient, who was informed that due to the rarity of this pathology, there was limited evidence available to guide management recommendations. Given his young age and the paucity of data to guide surveillance, our team recommended transanal minimally invasive surgery (TAMIS) to excise the lesion. However, the patient declined surgery at this time. He has been booked for a surveillance flexible sigmoidoscopy in 6 months.

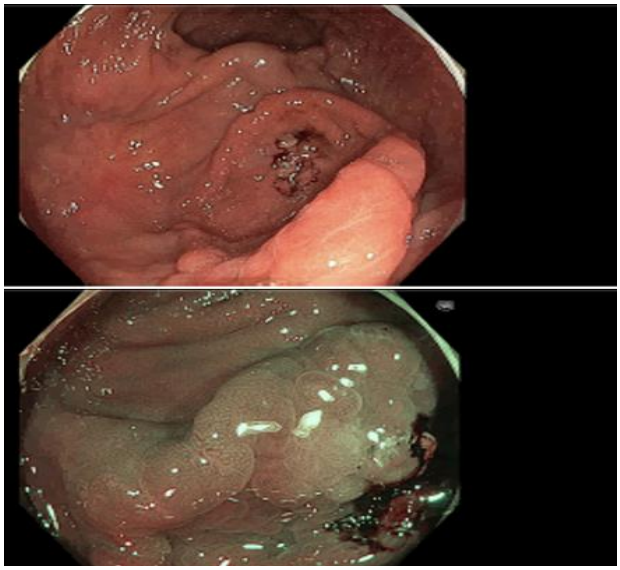

Figure 1: Endoscopic images of gastric heterotopia in the distal rectum.

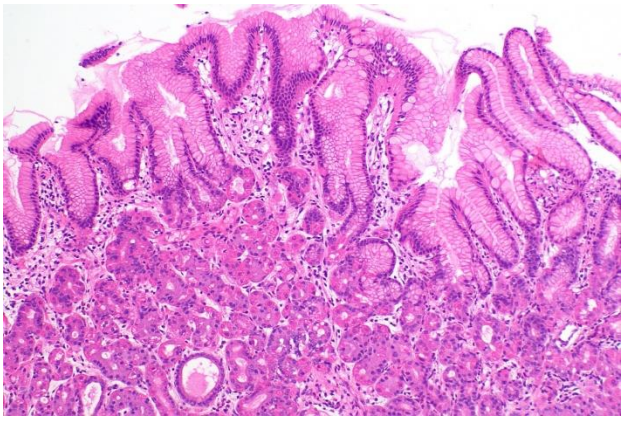

Figure 2: Rectal biopsy with hematoxylin and eosin Staining: oxyntic glands and parietal cells consistent with tissue usually found in the stomach.

DISCUSSION

GH can occur throughout the digestive tract and there are some reported cases in the mediastinum, biliary tract and scrotum.[4] GH in the rectum is rare.

The pathophysiology remains uncertain however multiple theories have been proposed to explain its occurrence. Congenital theory suggests GH arises from errors in cell positioning during descent of the stomach through embryogenesis.[5] Whilst this provides an explanation for GH in the oesophagus, it cannot account for hindgut heterotopia as in the case discussed. Acquired theory proposes that GH arises from metaplasia of pluripotent endodermal stem cells either spontaneously or as a result of injury and inflammation.[5] There is only one documented case of GH in the rectum associated with inflammatory bowel disease (IBD) otherwise there has not been cases reported where there is an obvious inflammatory trigger for rectal metaplasia to occur in these patients.[6]

Over the last decade, GH in the gastrointestinal tract has more often been detected incidentally during screening endoscopies in asymptomatic patients.[1,5] A limited number of patients with rectal GH are documented to have rectal bleeding or perianal pain.[5]

Endoscopic examination of GH is typically mass forming, which can easily be mistaken for malignancy.[5] Histopathological analysis is the only accurate means of diagnosis. Given its rarity, it's important to consider the possibility of mislabelled specimens, particularly in cases where an upper endoscopy has been performed on the same day.

The risk of malignant transformation remains uncertain. Mannan et al reported five cases of pyloric gland adenoma (PGA) arising from GH in the rectum.[5] Of these cases, 2 were found to have associated focal adenocarcinoma.[5] To the best of our knowledge, these are the only documented cases of adenocarcinoma arising from GH in the rectum, indicating a possible preneoplastic role of GH.

The recommended treatment of GH in the rectum is unclear. In symptomatic patients, proton pump inhibitors (PPIs) or histamine 2 receptor antagonists have been prescribed with success to reduce rectal bleeding.[7] Among the documented cases, surgical excision is frequently the preferred treatment or endoscopic mucosal resection (EMR) for smaller lesions.[1,5,7] Given that surgical excision is often the chosen approach for GH in the rectum, there is almost no data available to guide surveillance of these lesions.

CONCLUSION

GH is uncommon and its presence in the rectum is particularly rare. With increasing number of screening endoscopy procedures, more case report**s** are emerging in the literature. It is most frequently diagnosed incidentally in the asymptomatic patients who may be hesitant to proceed with surgical management, especially given its malignant potential remains unclear. Management recommendations and justification for surgery can therefore be challenging due to a limited understanding of the natural history of GH and the paucity of available literature to guide evidence-based practice.

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